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A Typical Case of Jeavons Syndrome with Intellectual Disability

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Abstract: ***Jeavons syndrome is a rare, idiopathic form of reflex epilepsy, with a childhood onset (2 to 14 yrs) and a symptom peak at 6-8 years of age. The hallmark features are eyelid myoclonia (EM) with or without absences, electroencephalography paroxysms and photosensitivity. Eyelid myoclonia consists of marked jerking of eyelids often with jerky upward deviation of eyeballs and retropulsion of head. This may be followed by mild impairment of consciousness (brief absences). The seizures are brief and characteristically precipitated by eyelid closure several times a day, in presence of bright unflickering light. The main diagnostic procedure is Video-EEG where young untreated patients show photoparoxysmal discharges induced by photic stimulation. The prevalence is unknown and genetic studies unremarkable, but Jeavons syndrome appears to represent around 7-8% of all idiophatic generalized epilepsies (IGEs) and has a slight female preponderance. Generalized tonic-clonic seizures occur in most patients, but the frequency of the seizures is low. Mental development is usually normal but mild to moderate intellectual deficit has been reported in some cases. Here we present a 14 yr old female, born out of consanguinous marriage, normal delivery and normal birth weight, with delayed milestones, taken to the department of paediatric neurology with complaints of eyelid fluttering more on exposure to sunlight, poor scholastic performance, behavioral issues including shouting spells, self-biting and frequent tantrums. On examination, moderate intellectual disability was present. During photic stimulation, patient had a clinical attack of eyelid fluttering and EEG showed frequent paroxysmal bursts of spike and slow waves. MRI brain showed normal study. Eyelid myoclonia is often missed or misdiagnosed as facial tics causing a delay in diagnosis and intervention. With early diagnosis and timely intervention frequency and severity of paroxysms can be significantly reduced.***

Keywords: Jeavons syndrome, intellectual disability, seizures, EEG, childhood

1. Introduction

Jeavons syndrome is a rare, idiopathic form of reflex epilepsy. It constitutes around 7-8% of all idiopathic generalized epilepsies (IGE's) with a slight female preponderance. The onset is usually in childhood (2 to 14 yrs age) with a symptom peak at 6-8 yrs of age.

The Hallmark features include eyelid Myoclonia (EM) with or without absences, EEG Paroxysms and Photosensitivity. The seizures are brief, characteristically precipitated by eyelid closure, in the presence of bright unflickering light.

The prevalence is unknown and genetic studies unremarkable. The diagnostic test of choice is Video EEG, where photoparoxysmal discharges can be seen with photo stimulation.

2. Case Presentation

History
The informant was the father, being reliable and adequate.

The patient was a 14yr old female born out of 2nd degree consanguinous marriage, taken to the department of paediatric neurology 3yrs back with complaints of:

- Eyelid fluttering, on exposure to sunlight, lasting more than 10 minutes, associated with impairment of consciousness, with 10 to 15 episodes per day.
- Poor scholastic performance (Went to school upto 7 yrs age. Cannot write letters, only scribbles)
- Behavioral issues including shouting spells, self biting and frequent tantrums
- Delay in speech

The child plays with peers, takes initiative to interact but does not understand rules of the game. Herself care is independent (toilet trained, bathes and dresses herself, eats food and does simple chores at home)

Patient was now referred to the Dept of Psychiatry for behavioral issues and disability certification.

Past History: First episode of fluttering of eyelids noticed at the age of 6 yrs.

Over the years the father noticed increase in frequency and only when he took the patient for a walk in the sunlight.
No H/O GTCS like seizure episodes.

Birth History: Normal delivery, immediate birth cry, birth weight-2.7kg.

On Examination:
IQ-44 (Moderate Intellectual Disability)
Tone: normal and bilaterally equal in upper and lower limbs
Power: 5/5 in all 4 limbs

Milestones:
Gross motor-walk with support at 3 yrs (currently can walk without support, run, climb up and down the stairs)

Speech-onset at 5yrs (currently responds to questions relevantly, but cannot form full sentences. Comprehension is intact)

Investigations: (In untreated patient)

EEG (1 Hr Telemetry)-During Photic stimulation at >6Hz, clinical attack of fluttering of eyelids, during which record

showed paroxysmal bursts of spike and slow wave discharges are seen bilaterally and synchronously with frontal pole and frontal predominance.

MRI BRAIN-Showed normal study.

Treatment:
Pt is currently on the following medications-Tab Levitiracetam 750mg BD, Tab Clonazepam 0.25mg BD, Syp Valproate 5ml TID, Tab Folic acid 2.5mg OD.

A probable diagnosis of Jeavon's syndrome with Moderate Intellectual Disability with Hyperactivity was made and the patient was started on suitable antiepileptics as mentioned above.

With good compliance, the patient has shown a decreased frequency and severity of eyelid myoclonias or paroxysms.

3. Discussion

Jeavon's syndrome has been under-diagnosed, with the eyelid myoclonia often missed or misdiagnosed as facial tics causing delay in diagnosis and treatment onset. It is a lifelong disorder with better prognosis in men than women, well controlled with antiepileptics.

-First line-Valproate monotherapy or with Clonazepam/ Levetiracetam (anti myoclonic, anti photosensitive)
-Second line-with Lamotrigine or Ethosuximide (based on main seizure type)

Mental development is usually normal but borderline or mild to moderate intellectual deficit has been reported in some cases. In the long term, inevitably Generalized tonic-clonic seizures occur, either spontaneous or induced by light, precipitated by sleep deprivation, alcohol, or inappropriate antiepileptic modifications. Over time, the photosensitivity may disappear in middle age, but the EM persists (more resistant to treatment, often without apparent absences or photosensitivity).

It can be differentiated from other photosensitive epilepsies as they are sensitive mainly to flickering light.

Avoid seizure precipitants such as light or hyperventilation, can use special glasses/ blue ZI lenses for photosensitivity.

The component of intellectual disability can be managed by developing a comprehensive multidisciplinary management plan including special education, speech therapy, occupational therapy and community services that provide social support and respite for the affected families.

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